Diagnosis of Autism Spectrum Disorder using modern AI Techniques

Synopsis

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Abstract

Autism Spectrum Disorder is a complex neuro-developmental disorder with high prevalence and life long in nature. ASD individuals face certain difficulties such as social interaction , communication, restricted interest, repetitive behaviors, sensory sensitivities and behavioral challenges. It is a spectrum of disorder and its symptoms vary from individual to another. Also, the symptom severity of ASD varies with an individual's age. It consists of mainly five types of disorder, that are: Asperger's syndrome, Rett's Syndrome, Childhood disintegrative disorder, Kanner's syndrome and pervasive development disorder. There is no availability of any pathological test for differentiating between ASD individuals from Healthy individuals. Thus, psychiatrists need to diagnose individuals by either observation or interviews over sessions which in turn makes their task tedious. Currently, various machine learning and deep learning and related methodologies have been developed which affirm detection of ASD and aiding psychiatrists in their diagnosis task. As ASD is a spectrum of disorder hence there is a lot of scope in improvement of the existing models. With the help of deep learning multiple layer processing can be performed to extract features from data. This research aims at developing an aiding model for in-depth diagnosis of ASD.

1 Introduction

ASD is a combination of different neuro-developmental disorders that has a strong impact on social interactions, communication and behavior and is often associated with sensory sensitivity[3]. This is called the "spectrum" because it includes an extensive range of symptoms and degrees of severity. People with ASD often possess unique strengths and challenges, and their experiences can vary widely[2]. Different learning modalities that are affected in ASD are visual, auditory, kinesthetic, tactile and sensory.
Earlier mainly five major subtypes of ASD were classified as follows:

1. **Asperger's Syndrome:** APD, is a broader category which ASD affects children easily and has normal symptoms cognitive and verbal skills. But these individuals showcase more difficulties with social skills compared to other types of ASD and they repeat the same activities and activities that bring them joy. Children may experience less emotion and less self-expression[12].
2. **Rett's Syndrome:** Rett syndrome is a genetic neuro-developmental disorder that occurs almost exclusively in girls and rarely seen in boys. This disorder results in severe impairments which have an impact on almost every characteristic of a child's life: the ability to speak freely, to walk, to eat and even to breathe. The hallmark of Rett syndrome is almost continuous, repetitive hand movements. Rett syndrome is usually diagnosed in children of the age range 6 to 18 months when they begin to skip developmental milestones or lose learned skills[15].
3. **Childhood disintegrative Disorder:** Childhood disintegrative disorder (CDD), also known as Heller's syndrome and disintegrative psychosis, is a rare condition characterized by late onset (>3 years of age) of developmental delays in language, social function, and motor skills[16].
4. **Kanner's Syndrome:** This type of autism is also known as classic autistic disorder. Symptoms can include difficulty communicating or understanding others, poor eye contact, and hypersensitivity to stimuli (smell, light, sound, taste, or touch).
5. **Pervasive Developmental Disorder:** PDD is a group of disorders characterized by difficulties in social interaction, verbal and nonverbal communication, imagination, and a low number of repetitive interests and activities[12].

It is estimated that around 1 in 100 children worldwide has autism. These estimates represent the mean and the reported incidence varies significantly between studies. However, the numbers are quite higher as compared by some well known studies. The widespread presence of autism in many low- and middle-income countries is unknown[16].

Some key characteristics of Autism Spectrum Disorder that include:

- Repetitive and Restricted Behaviors: They engage in iterative behaviors or activities. These may include repetitive movements such as waving or rocking the hand, inflexible adherence to routines, strong attachments to specific objects, and intense focus on narrow interests.

- Social Communication Challenges: ASD individuals may face difficulty understanding and using verbal and nonverbal communication, forming and maintaining relationships, interpreting facial expressions, gestures, and body language.

- Sensory Sensitivities: ASD individuals experience sensory sensitivities, they are over or under-sensitive to sensory stimuli such as light, sound, texture, taste or smell. This can lead to sensory overload or avoidance of certain environments.

- Individual Differences: The symptoms and severity of ASD vary greatly. Some have significant cognitive and language deficits, while others have average or above average intelligence and good language skills.

- Co-occurring disorders: They have co-occurring disorders such as Anxiety, Intellectual Disability, Attention Deficit Hyperactivity Disorder (ADHD), OCD and Depression.

- Strengths: They have unique strengths for example might excel in areas like math, music, art, memory, and attention to detail.

The diagnosis of ASD requires a comprehensive assessment by a healthcare professional. Early diagnosis and intermediation can greatly improve results by helping people develop essential skills for communication, social interaction, and daily living.

It is vital that each person with ASD is an individual with its own kind of characteristics and needs. While the challenges can be significant, ASD individuals also make important contributions to society and can live fulfilling lives with the right support and understanding from their families, communities and society at large.

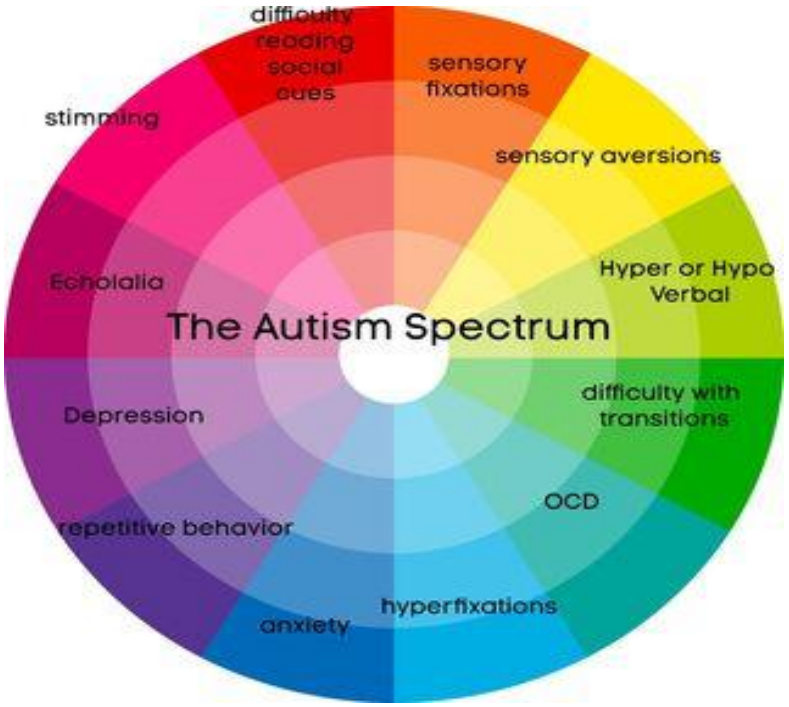

fig-1: Autism Spectrum Disorder[17]

Deep learning algorithms have proven to be advantageous when used in the diagnosis of ASD. ASD research involves analyzing diverse data sources such as behavioral assessments, neuroimaging data, genetic information, and more[1]. Deep learning algorithms can handle this multi-modal data and identify intricate patterns that may not be easily detectable using traditional methods. They can automatically extract relevant features from raw data, reducing

the need for manual feature engineering. This is particularly advantageous where identifying the right features can be challenging due to the disorder's heterogeneity. ASD detection requires capturing intricate and nonlinear relationships within the data. Deep learning models, particularly neural networks with multiple layers such as CNN, RNN, Attention based models can capture complex patterns and representations, potentially revealing subtle signals indicative of ASD. Also, various Computer vision techniques help in feature extraction tasks[2]. It can help reduce bias introduced by human judgment, as they learn from large datasets without inherent biases. This can lead to more objective and consistent detection outcomes. When applied to neuroimaging data such as (fMRI, sMRI, rs-fMRI) can uncover intricate brain patterns associated with ASD. This provides insights into the neural underpinnings of the disorder, helping researchers better understand how ASD affects brain structure and function. This encourages collaboration between computer scientists, neuroscientists, psychologists, and clinicians[10]. This interdisciplinary approach enriches the research process and enhances the quality of findings.

Major shortcomings of most research in the ASD realm has been the failure to take into account the heterogeneity of the ASD category

2 Literature Survey

In this section some previous research studies on ASD using Deep Learning are mentioned, which pioneer the background knowledge bases to proceed with related research work:

1. C. Su and et al (2020) [1] included description about DL algorithms that are best suited for particular areas of research in their paper- “Deep Learning in mental health outcome research: a scoping review”. They presented a list of research papers in the duration of 2014 to 2019 and grouped them according to type of data and their applications in certain areas such as clinical data, genetic data, vocal and visual expression data and social media data.
2. R. Anthony and et al (2020)[2] performed analysis of different datasets and methods for ASD in their paper- “Computer vision in autism spectrum disorder research: a systematic review of published studies from 2009 to 2019”. It states that deep learning approaches show superior performance for feature extraction and classification. Along with this multi-modal approaches provide even better results.
3. S. Raj and et al (2019)[3] trained various ML models such as Logistic Regression, SVM, Naive Bayes, KNN, ANN and CNN on ASD screening data available on UCI repository in their paper titled- “Analysis and detection of Autism spectrum disorder using machine learning techniques”. Here, CNN was the best performing model out of all other conventional models with an accuracy of 99.53%, 98.30% and 96.88% for each individual dataset. This paper worked on mainly pre-processing data and handling missing values.
4. N. Boughattas and et al (2022)[4] proposed ML algorithms based on detecting ASD individuals. In this paper- “Autism spectrum disorder detection using machine learning algorithm”. Here, 3 models were implemented: CNN, SVM along with PCA and LSTM on ABIDE (Autism Brain Imaging Data Exchange). The CNN model achieved accuracy of 95%, SVM+PCA achieved accuracy of 88% and LSTM achieved accuracy of 76.25%. They also stated that to achieve better recognition of ASD other modalities i.e EEG, speech or kinesthetic modalities can also be analyzed.
5. Z. Huang and et al (2020)[5] developed a GBFS (Graph based feature selection) method trained with DBN (Deep Belief Network Classifier). The paper- “Identifying Autism Spectrum Disorder from Resting – State fMRI using Deep Belief Network” was trained on ABIDE dataset with sample size 1035 entities. Their model successfully identified autistic neural correlation patterns from data driven outcomes with an accuracy of 76.4%.
6. M. Rakic and et al (2020)[6] presented a novel multimodal approach by combining sMRI and fMRI in the paper titled as- “Improving the detection of Autism spectrum disorder by combining structural and functional MRI information”. The ABIDE dataset-I was trained using MLP (Multi Layer Perceptron) which gave classification accuracy of

85.06%.

7. K. Vakadkar and et al (2021)[7] performed analysis of different classifiers such as LR(Logistic Regression), NB(Naïve Bayes), SVM (Support Vector Machine), KNN (K-Nearest Neighbor), RF (Random Forest) in the paper- “Detection of Autism spectrum disorder in children using machine learning techniques”. The Logistic Regression classifier performed best with accuracy of 97.15% on Q-CHAT (Quantitative Checklist for Autism in Toddlers). The limitation of this paper was availability of a large dataset.
8. I. Wiratsin and et al (2021)[8] presented a paper- “Feature Selection Technique for Autism Spectrum Disorder”. Here, ASD screening dataset was passed to the SVM (Support Vector Machine) classifier and Apriori algorithm was used to find association rules. This helped in extracting dominant features for designing ASD therapy programs.
9. Y. Zhang and et al (2022)[9] proposed an EEG metrics based prediction model for analyzing the intensity of autistic symptoms in the paper- “Predicting the symptom severity in Autism spectrum disorder based on EEG Metrics”. An EEG Dataset ABC-CT dataset-e NDAR has been used. This research found prominent relationships between Brain networks and symptoms of Autism. The outcome stated that a combination of EEG network and spectral metrics can help in improving prediction performance of ADOS Scores.
10. J. Han and et al (2022)[10] proposed a multimodal diagnostic approach by using a combination of EEG and ET (Eye Tracking) data. In their paper- “A multimodal approach for identifying Autism spectrum disorders in children '', a two step multimodal model was proposed based on SDAE (Stacked Denoising AutoEncoders) algorithm. First, two separate SDAE models were developed to learn EEG and ET functions, respectively. Subsequently, the third SDAE model was developed to perform multimodal fusion with concatenated features learned from EEG and ET. This model achieved 95.56% accuracy with optimized power detection. They also revealed future opportunities for the development of advanced neural network models and attention-based models to reduce the complexity of model training.
11. Y. Liang and et al (2021)[11] proposed a new CNNPL (CNN Prototype Learning) framework to classify brain functional connectivity for ASD in their paper- “A convolutional neural network combined with prototype learning framework for brain functional network classification of Autism spectrum disorder”. Here, the transfer learning approach was introduced to accelerate model training. CNN has been used to extract basic features from each of the prototypes by automatically prototype learning. Two cross validation schemes one is classical cross validation and second intersite cross validation has been performed to validate the model. The experimental results recognized the most discriminative ASD associated brain areas by analyzing features from top to bottom.
12. A. sadiq and et al (2022)[12] proposed a novel approach for extracting non-oscillatory connectivity from BOLD rs-fMRI in the paper- “Non- Oscillatory Connectivity Approach for classification of Autism Spectrum disorder subtypes using resting state fMRI”. The

performance for individual classification is recorded as 98.6%, 97.2% and (7.2% respectively and performance for combined multiclass classification is 88.9% on ABIDE-resting state fMRI (Functional MRI) data consisting of three equal size autism subtype data for ATD (Autistic Disorder), APD (Asperger's disorder), PDD (Pervasive Developmental Disorder- not otherwise specified) and healthy individuals. The outcome states that both individual classification and multiclass classification outperformed the Pearson's Coefficient Correlation model.

13. Ke. Fengkai and et al (2020)[13] proposed two models with separate datasets in their paper- "Exploring the structural and strategic bases of Autism spectrum disorders with deep learning". Their model successfully recognized different brain features contributing to classification. Here, RNN (Recurrent Neural Network) based RAM (Recurrent Attention model) achieved better results on child Psychiatric Clinic at Severance Hospital, (YUM)(NA publicly- licensed)) data and 3D CNN model achieved better results on ABIDE dataset.
14. S. M. Hasan and et al (2022)[14] in their paper- "A machine learning framework for early stage detection of Autism spectrum disorders" proposed best feature scaling techniques to improve classification performance. The feature scaling techniques used in this paper are: QT (Quantile Transformer), PT (Power Transformer), Normalizer and Max Abs scaler. The highest accuracies were achieved on Adaboost and LDA model while scaling Toddlers and Children with normalize and Adolescents and Adults with the QT method on ASD Screening dataset.

3 Objectives

Following are the major objectives of the research:

- Recognizing important features for distinguishing Autistic Individuals from Normal individuals.
- Identifying significant features in ASD subtypes.
- Design a Model to diagnose Autism spectrum Disorder individuals accurately.
- Performing Accuracy Evaluation of proposed model.

4 Methodology

After thorough study of currently proposed methods and their implementation, new research direction will be determined. This involves designing new strategies to extract prominent features. In our research we will be using Neural Network models. Firstly, we will go through different available datasets and find out the most effective features. Secondly, we will pre-process the dataset for removing ambiguity if any. Thirdly, this pre-processed dataset will be provided to train the proposed model. Finally, we will test our model and find its accuracy. We will also be focusing on finding out detailed insights and areas for further progress of research work.

5 Data resources

The grassroot of every research work starts with data available in that particular research work. Mainly in deep learning models your data acts as mainstream for your features. Thus, selection of proper and relevant data resources is very crucial in training any deep learning model.

Following are some helpful data resources for proceeding with Autistic research:

- ABIDE (Autism Brain Imaging Data Exchange): https://fcon_1000.projects.nitrc.org/indi/abide/.
- UCI Repository: ASD screening data for adult, children and adolescent https://archive.ics.uci.edu/datasets?search=Autistic%20Spectrum%20Disorder%20Screening%20Data%20for%20Children
- Quantitative Checklist for Autism in Toddlers(Q-CHAT): https://www.kaggle.com/fabdelja/autism-screening-for-toddlers
- EEG Dataset ABC-CT dataset-e National Database for Autism Research: NIMH Data Archive - NDA Home Page (nih.gov)

6 Social Relevance

The societal impact of ASD research is profound and far-reaching. Research on ASD is of great importance to people with ASD, their families, communities and society as a whole. Here are some key aspects that highlight the societal importance of ASD research:

- Economic Impact: ASD research has economic impact because it reduces the long-term costs associated with providing lifelong support to people with ASD and provides support to psychiatrists for diagnosis. Early interventions and effective treatments can lead to better outcomes, greater independence, and reduced dependency on social services.
- Global Impact: ASD is a global problem and research transcends geographic boundaries. International collaborative research promotes the sharing of knowledge, expertise and best practices, leading to a global understanding of autism and its treatment.

- Public Policy: ASD research plays a role in shaping public policies related to healthcare, education, employment, and social services. Informed policies can improve access to early interventions, therapies, and support services, making sure that people with ASD have the resources they need to thrive.
- Education and Advocacy: Research results form the basis for the development of evidence-based educational approaches and support systems. They help create inclusive classrooms, equip teachers with tools to meet diverse learning needs, and enable advocates to actively encourage policies which prove beneficial for ASD .

7 Proposed Plan

Year	Plan
1	Literature survey and basic implementation
2	Development of Detection model and formulate results.
3	Paper writing and publishing
4	Thesis writing

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